KRT81 gene

keratin 81

Normal Function

The *KRT81* gene provides instructions for making the type II hair keratin K81 protein (K81). This protein belongs to a group of proteins known as keratins, which are tough, fibrous proteins that form the structural framework of cells that make up the hair, skin, and nails. Each keratin protein partners with another keratin protein to form molecules called intermediate filaments. These filaments assemble into strong networks that provide strength and resiliency to the tissues and protect them from being damaged by everyday physical stresses. The K81 protein is found in cells that make up the inner compartment of the hair shaft known as the cortex, and this protein helps give hair its strength and elasticity.

Health Conditions Related to Genetic Changes

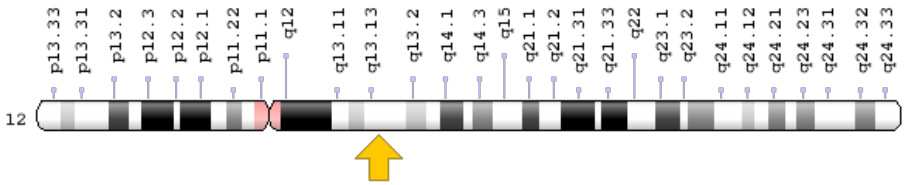
monilethrix

At least two mutations in the *KRT81* gene have been identified in people with monilethrix, a hair condition characterized by strands of hair with a beaded appearance and short, brittle hair that breaks easily. Mutations associated with this condition change a single protein building block (amino acid) in the K81 protein. The amino acid changes usually occur in a region of the K81 protein thought to be important in intermediate filament formation. In people with monilethrix, the cortex of the affected hair shaft appears abnormal. However, it is unclear how mutations in the *KRT81* gene are related to the abnormality in the cortex or the beaded appearance of the hair.

Chromosomal Location

Cytogenetic Location: 12q13.13, which is the long (q) arm of chromosome 12 at position 13.13

Molecular Location: base pairs 52,285,913 to 52,291,515 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ghHb1
- hair keratin K2.9
- hard keratin, type II, 1
- Hb-1
- HB1
- hHAKB2-1
- hHb1
- keratin-81
- keratin 81, type II
- keratin, hair, basic, 1
- KRT81_HUMAN
- KRTHB1
- type II hair keratin Hb1

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: Development and Structure of the Hair Follicle
  https://www.ncbi.nlm.nih.gov/books/NBK45997/#ch4829.s2

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28KRT81%5BTIAB%5D%29+OR+%28%28HB1%5BTIAB%5D%29+OR+%28Hb-1%5BTIAB%5D%29+OR+%28KRTHB1%5BTIAB%5D%29+OR+%28MLN137%5BTIAB%5D%29+OR+%28hHAKB2-1%5BTIAB%5D%29+OR+%28K81%5BTIAB%5D%29+OR+%28ghHb1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- KERATIN 81, TYPE II
  http://omim.org/entry/602153

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_KRT81.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=KRT81%5Bgene%5D
- HGNC Gene Family: Keratins, type II
  http://www.genenames.org/cgi-bin/genefamilies/set/609
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6458
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/3887
- UniProt
  http://www.uniprot.org/uniprot/Q14533

Sources for This Summary

- OMIM: KERATIN 81, TYPE II
  http://omim.org/entry/602153
- Korge BP, Hamm H, Jury CS, Traupe H, Irvine AD, Healy E, Birch-MacHin M, Rees JL, Messenger AG, Holmes SC, Parry DA, Munro CS. Identification of novel mutations in basic hair keratins hHb1 and hHb6 in monilethrix: implications for protein structure and clinical phenotype. J Invest Dermatol. 1999 Oct;113(4):607-12.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10504448
- Langbein L, Rogers MA, Winter H, Praetzel S, Schweizer J. The catalog of human hair keratins. II. Expression of the six type II members in the hair follicle and the combined catalog of human type I and II keratins. J Biol Chem. 2001 Sep 14;276(37):35123-32. Epub 2001 Jul 9.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11445569

- Rogers MA, Langbein L, Praetzel S, Moll I, Krieg T, Winter H, Schweizer J. Sequences and differential expression of three novel human type-II hair keratins. Differentiation. 1997 Feb;61(3): 187-94.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9084137
- Schweizer J, Bowden PE, Coulombe PA, Langbein L, Lane EB, Magin TM, Maltais L, Omary MB, Parry DA, Rogers MA, Wright MW. New consensus nomenclature for mammalian keratins. J Cell Biol. 2006 Jul 17;174(2):169-74. Epub 2006 Jul 10.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16831889
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2064177/

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https://ghr.nlm.nih.gov/gene/KRT81

Reviewed: March 2012
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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Department of Health & Human Services